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Aneurysmal rupture of the costo-cervical trunk in a patient with neurofibromatosis type 1: A case report

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ABSTRACT

INTRODUCTION: Rupture of blood vessels associated with neurofibromatosis type 1 (NF-1) is a rare but life threatening complication. We report the first case of an aneurysmal rupture from the costocervical trunk in a NF-1 patient treated by endovascular embolisation.
PRESENTATION OF CASE: A 43 year-old gentleman with a past medical history of NF-1 presented with sudden onset left sided neck swelling. A computed tomography (CT) revealed a large cervical haematoma, which was causing airway compromise, requiring the patient to be intubated. Percutaneous embolisation of the bleeding vessel from the costo-cervical trunk was performed with successful haemostasis and no immediate complications. A repeat CT scan showed a reduction in the original cervical haematoma. However, six days post embolisation, the patient arrested with complete whiteout of the left hemithorax.
DISCUSSION: CT angiography is the gold standard for diagnosis of an aneurysmal rupture in NF-1 patients, and percutaneous embolisation is the preferred modality in patients who are haemodynamically stable due to arterial fragility and high intra operative mortality rates. The increasing haemothorax could be explained by the original cervical haematoma draining down into the pleural space, or the possibility of a new second bleed.
CONCLUSION: This is the first reported episode of bleeding from the costocervical trunk in NF-1 patients. Ruptured aneurysms require urgent CT angiography, if haemodynamically stable, and further input from the vascular surgeons and vascular radiologists.

1. Introduction

Vascular involvement is rarely seen in NF-1 patients with an incidence of just 3.6%,[1] involving mainly larger blood vessels such as the subclavian artery. Acute vascular presentations associated with NF-1 are life threatening and require immediate attention.

We report a case of a massive left sided neck swelling caused by the rupture of an aneurysm from the left costocervical branch. An extensive cervical haematoma was formed, causing significant airway compression in a patient who already had major cervical spine abnormalities. The rupture was treated endovascularly with microcoils.

2. Presentation of case

We present a case of a 43 year-old gentleman, with a background history of neurofibromatosis type 1 with plexiform neurofibromas who awoke with sudden onset left sided chest pain and a progressive left sided neck swelling. On admission, the patient's vital observations were stable. Haemoglobin and haematocrit level were 14.1 g/dL and 0.41 L/L, respectively, and clotting screen was normal. A chest radiograph demonstrated tracheal deviation to the right side, a widened mediastinum and a left sided pleural effusion. A Computed Tomography (CT) scan of the neck revealed an extensive left sided cervical haematoma (Fig. 1), causing significant airway compression, requiring intubation. A CT angiogram demonstrated a bleed from the left costocervical trunk which had also spread into the ipsilateral pleural space, causing a small haemothorax (there was no other vascular malformation identified on the CT scan to account for the haemothorax). The patient was immediately transferred to a vascular centre where a digital subtraction angiogram was performed via the left brachial artery approach. The bleeding vessel from the lateral branch of the left costo-cervical trunk was identified and appeared to be a ruptured pseudo-aneurysm. One of the branch arteries distal to the site of bleeding was successfully negotiated and both the outflow and the inflow were satisfactorily embolised with multiple microcoils (Tornado Platinum Microcoils, Cook Medical Inc., Bloomington, IN) varying in size from 3 to 5 mm (Fig. 2). There were no immediate complications (Fig. 3) and the patient's haemoglobin level was stable at 10.4 g/dL (haematocrit of 0.32 L/L) in the immediate post procedure period. He was then transferred to the intensive trauma unit.

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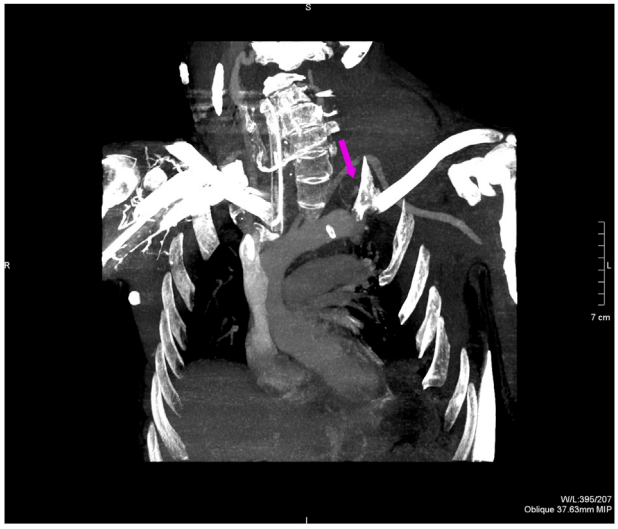

Fig. 1. Coronal reconstruction showing the site of the cervical haematoma (arrow). This is the original CT scan on admission. There is no 'whiteout' of the left lung on admission.

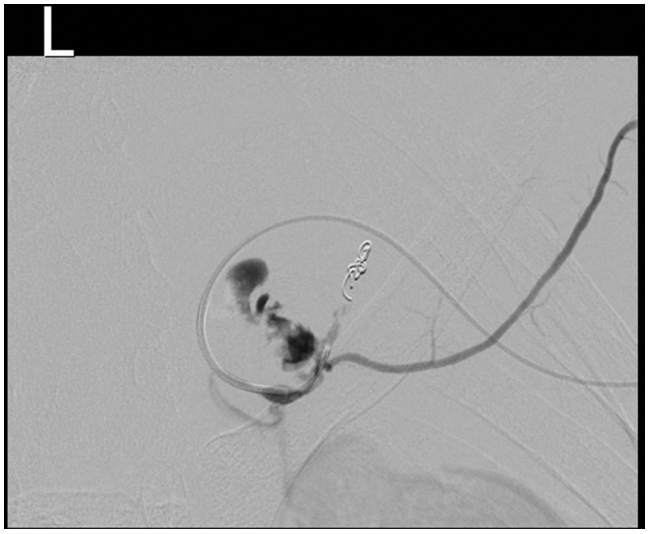

Fig. 2. Embolisation at the site of bleeding.

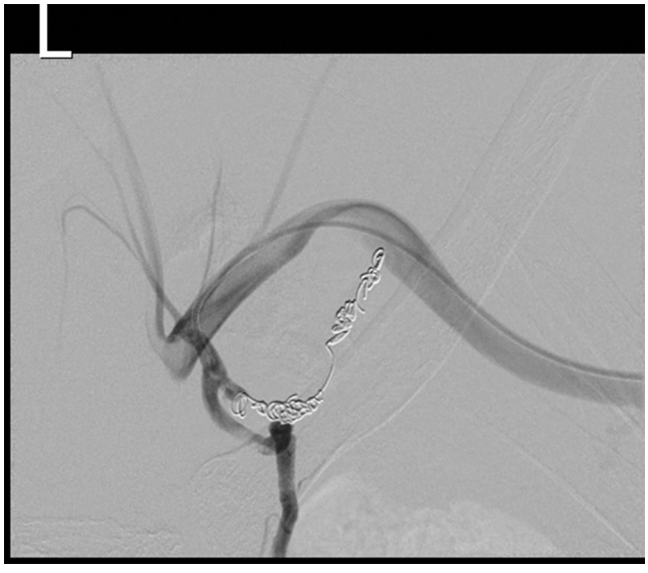

Fig. 3. Post embolisation demonstrating haemostasis and no active leak.

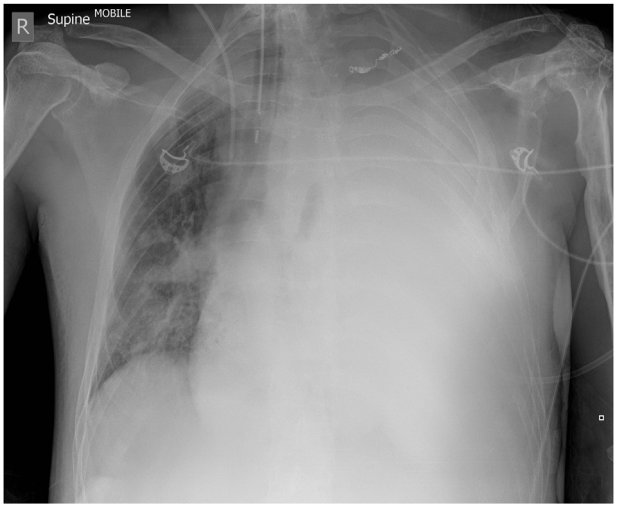

Fig. 4. CXR demonstrating complete 'whiteout' of the left hemithorax.

A repeat CT scan of the neck and thorax performed four days post embolisation confirmed no evidence of further bleeding in the neck and a reduction in the size of the original left cervical haematoma; now 10 cm × 8 cm × 7 cm. The size of the left haemothorax however had increased. Extensive pulmonary embolisms were also incidentally noted and a caval filter (Cook Celect IVC filter) was placed in the infra-renal IVC, via the right internal jugular vein approach. The patient was not on pharmacological venous thromboembolism prophylaxis due to the haemorrhage but was wearing stockings in addition to intermittent pneumatic compression. In light of the CT results, therapeutic low molecular weight heparin was commenced. Whilst awaiting transfer for possible thoracic decortication at a cardiothoracic centre, a central venous catheter (CVC) was inserted via the right internal jugular vein approach for inotropic support during transfer. Upon positioning of the CVC, the patient's systolic blood pressure immediately dropped to 50 mmHg and heart rate increased to 150 beats per minute. A chest radiograph showed a very large haemothorax with complete whiteout of the left hemithorax (Fig. 4). The patient quickly lost cardiac output and cardio-pulmonary resuscitation (CPR) was commenced. Thoracic drainage of the left haemothorax was then performed, draining 2.5 L of blood. Five units of blood were transfused during CPR but the patient did not survive. Haemoglobin and haematocrit levels were 6.4 g/dL and 0.19 L/L, respectively, at the time of cardiac arrest. Autopsy revealed the direct cause of death to be a soft tissue haemorrhage.

3. Discussion

Neurofibromatosis type 1 (NF-1) is an autosomal neurocutaneous disorder with a prevalence of about 1 per 3000 births, caused by a mutation of a gene on the long arm of chromosome 17 which encodes the protein neurofibronin.[2] It is typically characterised by café au lait spots, intertriginous freckling and neurofibromas.[3] Vascular abnormalities are rare (affecting just 3.6% of NF-1 patients) and comprise of stenoses, aneurysms, occlusions and arteriovenous malformations, mainly of the larger blood vessels.[1]

There have been many attempts to describe the association between neurofibromatosis and its vascular abnormalities. Greene et al. hypothesised that larger arteries such as the aorta, subclavian, carotid and proximal renal arteries are surrounded by neurofibromatous tissue (schwannoma, neurofibroma, or neurofibrosarcoma) resulting in proliferation of the intima, thinning of the media and fragmentation of the elastic layer, subsequently causing stenosis or

Fig. 5. Small size of the trachea (red arrow) and hence difficult insertion of endotracheal tube. Yellow arrow indicates large left cervical haematoma. (For interpretation of the references to color in text, the reader is referred to the web version of this article.)

aneurysm formation.[4] Leier et al. proposed that neurofibromatous tissue compresses the vasa vasorum of large arteries, weakening the segment of the artery secondary to ischaemia.[5] Ishizu et al. carried out histological examinations of a neurofibromatosis patient with a ruptured aneurysm and reported intimal thickening by proliferation of fibromuscular cells as well as media irregularity.[6]

The current literature contains reports on ruptures from the subclavian artery, intercostal, vertebral and lumbar arteries, in addition to a couple of reports of bleeding from the thyro-cervical trunk.[1] To the best of our knowledge, this is the first report of a rupture from the left costo-cervical branch to be associated with neurofibromatosis. The autopsy result leads us to believe that there may have been a second site of bleeding that was not originally identified on the CT scan or that there was a further bleed from the original site which drained into the left pleural space.

CT angiography is considered the gold standard in the diagnosis of ruptured aneursysms.[7] To provide symptomatic relief, thoracic drainage tubes can be inserted when ruptures cause a significant haemothorax. Our patient had multiple chest wall deformities, including acute severe kyphosis with vertebral body deformities of cervical spine C4–C7. The nature of the skeletal deformities would have made thoracic drainage tube insertion potentially hazardous increasing the likelihood of potential damage of intercostal arteries. His skeletal abnormalities also caused extreme difficulty during insertion of his endotracheal tube (Fig. 5) and he remained intubated during hospital stay.

When treating a ruptured aneurysm, endovascular therapy (involving coil embolisation or stent graft placement) has begun to replace surgical ligation of the bleeding vessel as the preferred modality[1,3,5,7–12] as it is less invasive and surgical repair is considered difficult due to arterial fragility in neurofibromatosis. Furthermore, due to the patient's extensive chest wall deformities, an endovascular approach was considered more favourable. Ultimately the modality of treatment depends on haemodynamic stability, with endovascular intervention tending to be the preferred method in haemodynamically stable patients (as in our case).[12] Some patients have been treated with thoracic drainage alone for symptomatic relief.[12] However in this case, since our patient's airway was compromised, it was imperative that the bleed from the ruptured vessel was stopped.

Despite the outcome in this situation, we feel endovascular intervention was still successful as the bleeding was immediately stopped, with the second CT scan also confirming the original haematoma to be reduced four days after embolisation. There has been one other case of a death in a patient treated with coil embolisation whereby a ruptured ascending cervical artery aneurysm was well embolised but, shortly after the embolisation, fatal haemorrhage was induced by dissection of the vertebral artery. On the other hand, a review by Fedoruk et al. found that surgical operative mortality rates in ruptured neurofibromatosis vessels are as high as 33%.[12]

4. Conclusion

Vascular complications in NF-1 patients must be recognised immediately and intervention performed quickly to prevent further deterioration. Endovascular treatment with coil embolisation remains the preferred modality of treatment when patients are haemodynamically stable. Vascular complications associated with NF-1 are difficult to manage due to the rarity of NF-1 itself and the infrequent occurrence of spontaneous aneurysmal ruptures. Extensive skeletal deformities add to the complexity of these cases. Optimal patient care requires a strong multi-disciplinary team, with input from vascular surgeons and vascular radiologists. Access to critical care unit and cardiothoracic backup are vital.

Conflict of interest statement

None declared.

Funding

None declared.

Ethical approval

Written informed consent was obtained from the patient's next of kin, as the patient had passed away, for publication of this case report and accompanying images.

A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Authors' contribution

Bhupinder Hoonjan – designing and writing the whole case report, analysis of the literature review, collecting patient information and images. Nagendra Thayur – preparation and reporting of images, assistance in writing the article, final approval for manuscript submission. Abdusalam Abu-Own – final approval for manuscript submission and assistance with writing the article.

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